MBD5 gene

methyl-CpG binding domain protein 5

Normal Function

The *MBD5* gene is one of a family of genes called the methyl-CpG-binding domain (MBD) genes. These genes provide instructions for making proteins that help regulate gene activity (expression) by modifying chromatin, the complex of DNA and protein that packages DNA into chromosomes. The MBD5 protein is likely involved in regulating gene expression and controlling the production of proteins that are involved in neurological functions such as learning, memory, and behavior. The MBD5 protein also seems to play a role in the growth and division (proliferation) and maturation (differentiation) of various types of cells.

Health Conditions Related to Genetic Changes

MBD5-associated neurodevelopmental disorder

Mutations in the *MBD5* gene have been found to cause *MBD5*-associated neurodevelopmental disorder (MAND). MAND is a condition that affects neurological and physical development from birth. Affected individuals often have intellectual disability, developmental delay, impaired speech, sleep problems, distinctive facial features, and mild hand and foot abnormalities. Most people with MAND also have behavior problems similar to autism spectrum disorder, a developmental condition that affects communication and social interaction.

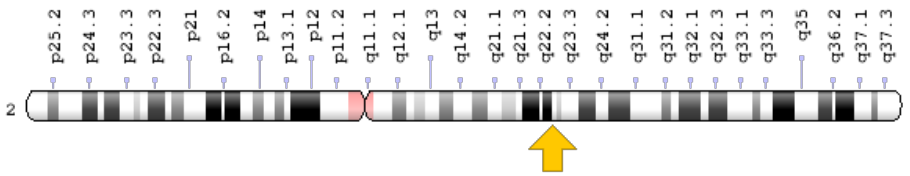
Some *MBD5* gene mutations that cause MAND delete large segments of the *MBD5* gene or result in the production of an altered protein with no function. These mutations affect one copy of the gene in each cell. As a result, there is less MBD5 protein available to regulate the expression of certain genes, leading to uncontrolled protein production. Proteins that play a role in neurological functions are particularly affected, which helps explain why MAND impacts brain development and behavior.

Additionally, an extra copy of the *MBD5* gene or a loss of the whole gene in each cell can cause MAND. In these cases, MAND results from abnormal copying (duplication) or removal (deletion) of a small piece of the long (q) arm of chromosome 2 at position 2q23.1. The duplicated or deleted segments can vary in size but always include the *MBD5* gene, and often additional genes. Researchers believe that *MBD5* gene changes underlie most of the signs and symptoms of MAND. Features of the condition generally do not differ based on the genetic cause, although they can vary between individuals.

Chromosomal Location

Cytogenetic Location: 2q23.1, which is the long (q) arm of chromosome 2 at position 23.1

Molecular Location: base pairs 148,021,011 to 148,516,971 on chromosome 2 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- methyl-CpG-binding domain protein 5
- methyl-CpG-binding protein MBD5

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database: Chromatin Mechanisms Regulating Gene Expression In Health And Disease
  https://www.ncbi.nlm.nih.gov/books/NBK45032/

Clinical Information from GeneReviews

- MBD5 Haploinsufficiency
  https://www.ncbi.nlm.nih.gov/books/NBK390803

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28MBD5%5BTIAB%5D%29+OR+%28methyl-CpG+binding+domain+protein+5%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- METHYL-CpG-BINDING DOMAIN PROTEIN 5
  http://omim.org/entry/611472

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=MBD5%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:20444
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:55777
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/55777
- UniProt
  https://www.uniprot.org/uniprot/Q9P267

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